



HSD3B2 gene

hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 2

Normal Function

The *HSD3B2* gene provides instructions for making the 3-beta-hydroxysteroid dehydrogenase (3 β -HSD) enzyme. This enzyme is found in the gonads, which are the ovaries in females and testes in males, and in the adrenal glands, which are located on top of the kidneys. Within these hormone-producing tissues, the 3 β -HSD enzyme is necessary for the production of many hormones, including cortisol, aldosterone, androgens, and estrogen. Cortisol has numerous functions such as maintaining energy and blood sugar levels, protecting the body from stress, and suppressing inflammation. Aldosterone is sometimes called the salt-retaining hormone because it regulates the amount of salt retained by the kidney. The retention of salt affects fluid levels and blood pressure. Androgens and estrogen are essential for normal sexual development and reproduction.

Health Conditions Related to Genetic Changes

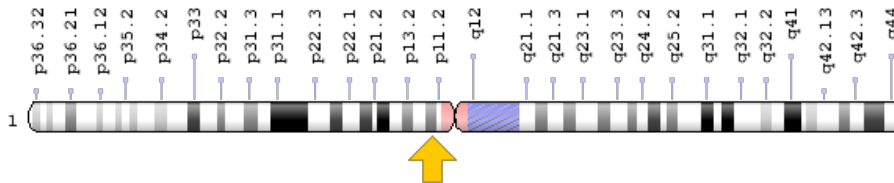
3-beta-hydroxysteroid dehydrogenase deficiency

At least 37 mutations in the *HSD3B2* gene have been found to cause 3 β -HSD deficiency. Most of these mutations change single protein building blocks (amino acids) in the 3 β -HSD enzyme, which typically reduces the activity of the enzyme. Mutations that allow the production of some functional enzyme, although at reduced levels, cause the less severe, non-salt-wasting or non-classic forms of 3 β -HSD deficiency. Other mutations result in the production of an abnormally short, completely nonfunctional 3 β -HSD enzyme, which causes the more severe, salt-wasting form of this condition. All types of 3 β -HSD deficiency interfere with the production of a variety of hormones and lead to abnormalities of sexual development and maturation.

Chromosomal Location

Cytogenetic Location: 1p12, which is the short (p) arm of chromosome 1 at position 12

Molecular Location: base pairs 119,414,931 to 119,423,039 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 3 beta-HSD type II
- 3-beta-hydroxy-5-ene steroid dehydrogenase
- 3-beta-hydroxy-delta(5)-steroid dehydrogenase
- 3 beta-hydroxysteroid dehydrogenase 2
- 3 beta-hydroxysteroid dehydrogenase type II, delta 5-delta 4-isomerase type II, 3 beta-HSD type II
- 3 beta-ol dehydrogenase
- 3BHS2_HUMAN
- delta 5-delta 4-isomerase type II
- HSD3B
- HSDB

Additional Information & Resources

Educational Resources

- Endocrinology: An Integrated Approach (first edition, 2001): The Major Steroidogenic Pathway in the Human Testis
<https://www.ncbi.nlm.nih.gov/books/NBK29/?rendertype=box&id=A1174>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28HSD3B2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 3-BETA-HYDROXYSTEROID DEHYDROGENASE 2 DEFICIENCY
<http://omim.org/entry/201810>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_HSD3B2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=HSD3B2%5Bgene%5D>
- HGNC Gene Family: Short chain dehydrogenase/reductase superfamily
<http://www.genenames.org/cgi-bin/genefamilies/set/743>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=5218
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3284>
- UniProt
<http://www.uniprot.org/uniprot/P26439>

Sources for This Summary

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